The HCU Herald

Featuring...

HCU Hero

Chris from Massachusetts

November 2024

All things Homocystinuria: patient stories, resources, research, events and more!

HCU HERO: CHRIS FROM MASSACHUSETTS

When I met my husband Chris 8 years ago, he was 28 years old, and he told me he had a blood clotting condition called Antithrombin 3 deficiency with elevated homocysteine levels and a MTHFR mutation. I didn't question it at first, but with a clinical background, after a few years, I knew something was off.

Chris had his first DVT blood clot at age 14. By his early 20's he had a massive stroke that crossed the midline after his care was delayed and the advice for a thrombectomy (a surgical procedure that removes a blood clot, or thrombus, from an artery or vein) was ignored. While waiting for medical attention, he was left in a hallway bed and had multiple drug tests run (during a period of time that spanned over 2 shift changes!) because they wanted to first ensure that he wasn't 'drug-seeking'. By the time his third drug test was negative, his clot turned to a bleed and crossed the midline covering 60% of his brain. Around the same time, he had a severed carotid artery with 80% stenosis (stenosis is a narrowing that can lead to restricted blood flow or pressure on surrounding tissues).

As the medical system failed to recognize his symptoms as manifestations of a bigger issue at play, Chris went on to experience even more symptoms: vision issues, multiple DVTs, venous stasis ulcers (open sores that occur due to poor blood circulation in the veins of the legs), and severe anxiety. He was diagnosed with bipolar disorder and had mental health medications shoved at him, even though nothing worked, and the medications magnified things.

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I started to dig deeper into Chris's medical records. He had seen the same provider from birth up to adulthood for primary care. He had been with the same hematologist since he was 14. None of his medical history added up; he only had 3 previous homocysteine levels checked, all of which were in the 300-400's, and he had been on B12 and folic acid for nearly 20 years at the time- with no reduction in his levels. I spent over a year researching and delving into things...I spoke with specialists and asked why they would tell him a homocysteine level in upper 300's is acceptable because it was lower than the 400's that he'd previously had. Dead end after dead end.

Two years ago, I left the clinical world and moved into my dream job at Harvard University working in Molecular and Cellular Biology with the research labs. This provided me additional time and knowledge to dig further. Earlier this year, I became convinced Chris had homocystinuria but couldn't get any of his medical providers to listen and they just kept saying "he is treated for elevated homocysteine levels, or "homocysteinemia". Nowhere in his medical records did I see that a genetic test to rule out a CBS-deficiency (classical homocystinuria), was performed, and I also discovered he didn't actually have a mutation of MTHFR, but rather two copies instead which was not the leading factor in his homocysteine elevation. I couldn't find that even one amino acid panel had been performed, not one urine amino acid or urine homocysteine level... NOTHING. By this point, Chris had suffered a massive stroke, experienced multiple DVTs, had two retinal intraocular lens surgeries, and webbed lungs, liver, and kidneys. I knew that I couldn't wait to take further action, and by April 2024 I was prepared to present his hematologist of 22 years with my research and demand answers!

I accompanied Chris to his hematology appointment and politely and professionally asked his provider all of the questions I had prepared. She knew immediately that I knew what I was talking about, and that she had been wrong. And she needed to know that I wasn't going to let this go. I insisted to her that Chris had a metabolic condition and that I knew that she knew that. I reminded her that it is not her area

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of specialty and questioned why she never referred him to genetics or metabolics. I also explained to her the difference between having 2 copies of a specific MTHFR mutation (C667T) and having classical HCU, and how more frequent blood levels and genetic testing could have helped surface this earlier. She had no real answers, and all her clinic notes spoke of was his mental health condition of bipolar, which we now know he doesn't even have. I told her I wanted him to take betaine, and she indicated that she'd never heard of it. Finally, I told her that if she would just give us the referrals to genetics/metabolics, we would leave. She said nothing other than calling me rude again, but she put the referrals in!

Here's a timeline of events that took place shortly after that:

- May: we met with pediatric genetics, and they performed a genetic test. She told me not to start him on Betaine, B6 or anything else; that we would wait to see the results.
- June: I didn't listen. My gut told me that Chris couldn't wait to start these medications. Chris began taking Betaine and B6, continued B12 and Folic Acid, and we added in NAC, and Deplin. We reduced his protein to 60g or less/day and changed all dairy to Almond/Soy or lactose-free products.
- July: The genetic test showed that Chris did indeed have CBS deficiency (Classical HCU). I wept. And wept.
- July 25: We went to a metabolic specialist in Boston where the diagnosis was confirmed. The specialist said she couldn't believe I had started him on the necessary medications already and she was impressed. They redrew his labs, and after the longest 24 hours of my life, the results were in! Chris went from an initial homocysteine level of 450.6 down to 38.9 since implementing medications and making dietary modifications on our own. I have NEVER cried so hard in my life. It wasn't even the validation of being right, but more so the knowing that there was HOPE!

Chris has finally been referred to many new specialists at the new hospital. Here we are, 22 years later. We've finally got a diagnosis of classical homocystinuria- and we are feeling hopeful!

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We've been married for two years now. Chris can play a guitar like no other, writes beautiful letters to me that heal my heart, works heard to learn what he doesn't know, cleans better than I do, folds laundry and does dishes and cuts grass. He loves to travel, hike, take pictures, go to concerts, and collects vinyl records! And, he cares for my children as if they were his own. My 15-year-old has chronic medical issues, and Chris flushes his PICC line and cares for him as if nothing were wrong with him at all. So, while Chris may have trouble understanding social situations, because the bipolar wasn't bipolar, but was Autism Spectrum...and his learning disabilities in school weren't defiance, but an untreated metabolic condition. And while past girlfriends may not have understood him and gave up, I'm so thankful we found each other and I have been able to get us here right now, to a homocysteine of 38.9.. and to be loved endlessly by this man who gets me, and I get him. Well, I'd say it doesn't get much better than this, other than seeing that homocysteine level drop into normal range, one day soon!

I'll end by saying that finding HCU Network America was huge for us. After receiving the diagnosis, Chris wanted to know that there were others out there; a community of support. Through a Google search, I found information about HCU Network America's Family Conference from back in June. From there, I connected with the Support group on Facebook and soon found the community that we were looking for. I feel it means that he's not alone...Living your whole life trying to make sense of things, when you finally get a diagnosis, knowing that although you're rare, having a community that "gets it" changes your whole outlook. When Chris saw that there are people like him out there, it sparked a motivation to share his story and to advocate, so that no one else has to go through what he's been through.

Click here to read Chris's & other patient stories on our website!

HCU AWARENESS MONTH – ADVOCACAY IN ACTION!

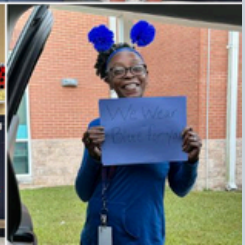
National White Cane Awareness Month

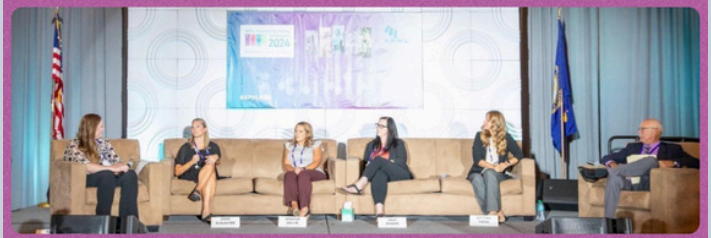
Meet Mason!

We are honored to celebrate and recognize our very own 4th grader, Mason, who takes pride in raising awareness for National White Cane Awareness Month. As a student with a visual impairement, Mason wants our Rosehill family to know that National White Cane Safety Day is recognized on October 15th.

National White Cane Safety Day annually celebrates the achievements of people who are blind or visually impaired. The day marks the importance of recognizing the white cane. The white cane is not only a tool. It also represents the independence of those who are blind worldwide.

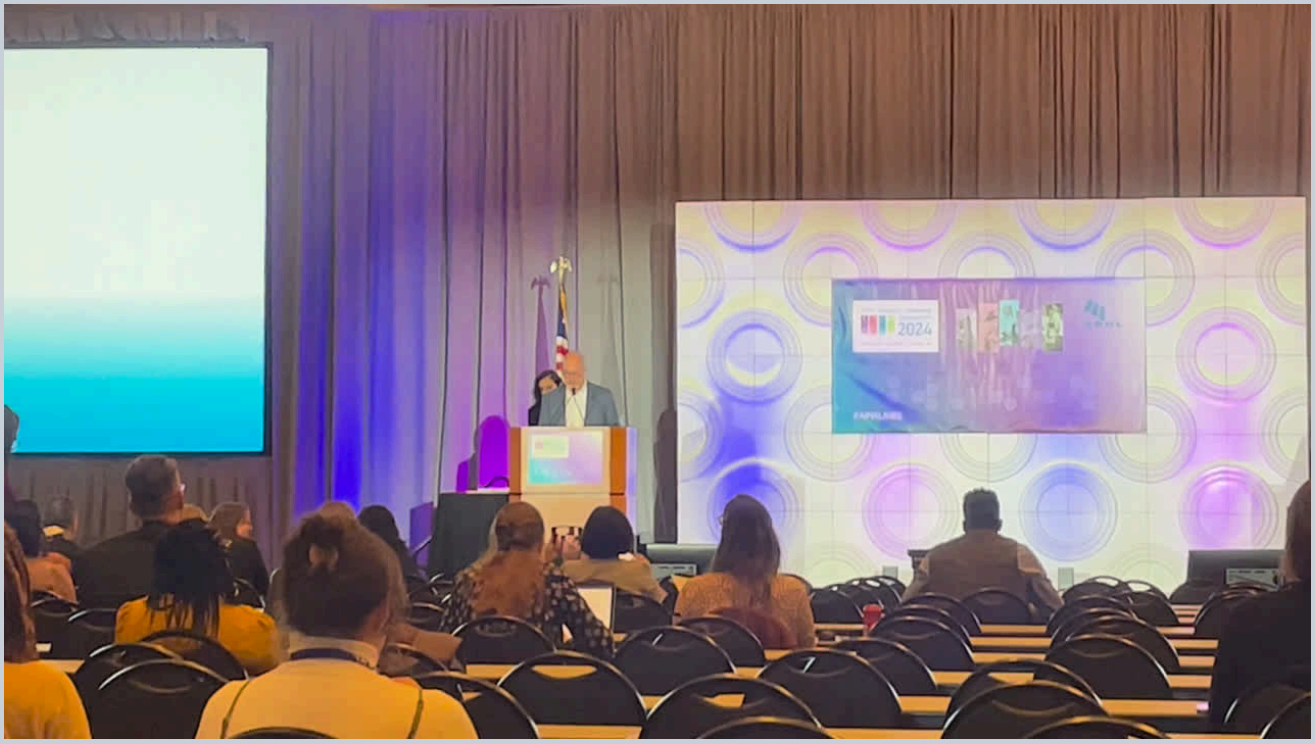
We commend and celebrate Mason for his dedication to bringing awareness to White Can Safety Day and we ask that you share this information with your children. If they see Mason in the halls please make sure they say hello and ask to learn more about his white cane!





NEWBORN SCREENING

Dr. Carla Cuthbert (CDC) receives Harry Hannon Laboratory Improvement Award



Carla Cuthbert, Chief of the Newborn Screening and Molecular Biology lab (CDC), was awarded the prestigious Harry Hannon Laboratory Improvement Award at the APHL (*Association of Public Health Laboratories)* Newborn Screening Symposium October 20-24, 2024 in Omaha, NE.

This award honors a person working in newborn screening to have made significant contributions in one or more of the following areas: assuring the quality of testing, enhancing the specificity of tests, establishing new creative laboratory approaches and technologies, providing laboratory training/education for new technologies and tests, or improving the detection of newborn disorders/conditions. The contributions of the recipient have had a direct effect in improving the quality of laboratory results for the newborn screening system.

HCU Network America would like to congratulate and thank Dr. Cuthbert and her team for their incredible contributions to improving Newborn Screening, particularly their work in developing an improved methodology for the newborn screening for homocystinuria.

To learn more about Carla and her team's development of a 1st-tier multiplex approach using total homocysteine, click **here**

Raising Joy

Double Good

Give Good. Get Good. That's Double Good!

When you buy Double Good's award-winning premium popcorn, 50% of your purchase goes to support HCU Network America!

100% contactless!
The product ships directly to the buyer.

To create your own pop-up store:
1) Download the Double Good App
2) Enter our event code: FKXZCA
3) Tap 'my store' to create & personalize your pop-up store

11.07.24 @ 3 pm to 11.11.24 @ 3 pm

As an added bonus, **all our proceeds will be doubled** as part of our end-of-year match!

HCU NETWORK AMERICA

MEET CHRIS AND HOLLY

a story of hope in the face of misdiagnosis

“Serious, but treatable”

is a phrase often used to describe conditions like homocystinuria (HCU). When identified early, HCU can be managed with treatment, allowing individuals to live typical lives. Unfortunately, approximately 50% of patients are missed by newborn screening, resulting in delayed diagnoses and sometimes life-threatening consequences. Two of the most common symptoms of untreated homocystinuria are blood clots and strokes. Individuals with these symptoms are often first seen by ophthalmologists and hematologists, where the correct diagnosis of homocystinuria may be missed.

Today, we want to share a story that embodies resilience, determination, hope, and the profound impact of a misdiagnosis — Chris and Holly’s story.

When Holly met Chris eight years ago, she quickly learned about his complex medical history. Chris had been diagnosed by a hematologist with a blood clotting disorder and elevated homocysteine levels.

Chris’s journey began at the age of 14 with his first deep vein thrombosis (DVT). By his early 20s, he had suffered a massive stroke due to delayed medical care and misdiagnosis. This stroke left him with significant brain damage, vision issues, and a host of other health problems.

HCU NETWORK AMERICA

Over the years, Chris faced multiple DVTs, venous stasis ulcers (non-healing open sores on the lower leg), and severe anxiety, all while being misdiagnosed with bipolar disorder.

Holly, with her clinical background, knew something was amiss. She spent years researching and advocating for Chris, confronting specialists and demanding better care. Her persistence paid off when she finally got Chris the correct diagnosis: classical homocystinuria. This rare metabolic disorder had been the root cause of his elevated homocysteine levels and numerous health issues.

With an accurate diagnosis, Holly's unwavering advocacy resulted in the introduction of new medications and dietary changes that reduced Chris's homocysteine levels from a life-threatening 450 to a manageable 39. He no longer needed numerous psychiatric medications and began feeling like a completely new person, as if a heavy fog had lifted. This transformation has given both Chris and Holly hope for a healthier future.

Chris's story is a testament to the importance of accurate diagnosis and the power of advocacy. It highlights the critical need for continued research, education, and support for those with homocystinuria.

HCU Network America acknowledges the significant gap in education within the field of hematology which can

lead to misdiagnoses for patients with homocystinuria. To address this, we have developed a Hematologist Toolkit designed to assist hematologists in identifying the symptoms of undiagnosed homocystinuria and facilitate referrals to genetic specialists.

We are also excited to announce that HCU Network America will be participating in the American Society of Hematology's (ASH) 66th Annual Meeting and Exposition in San Diego, CA. Our primary objective at this event is to bring awareness to hematologists and educate physicians on recognizing the signs and symptoms of homocystinuria, as well as to offer guidance on patient referrals to genetic experts. We are looking forward to building relationships with the over 30,000 attendees from over 100 countries in attendance! Additionally, we are in the process of creating a guidebook specifically for patients diagnosed in adulthood or later in life, to help them navigate the complexities of the medical system, and to educate them on the treatment for HCU.

As we approach the end of the year, we kindly request your support to continue our mission. Your generous donations enable us to provide essential resources, support, and hope to families like Chris and Holly's. Together, we can make a meaningful difference in the lives of those affected by homocystinuria.

“Chris's story is a testament to the importance of accurate diagnosis and the power of advocacy. It highlights the critical need for continued research, education, and support for those with homocystinuria.”

Matching Gift Challenge

Three generous donors have pledged to match EVERY gift up to $25,000 of donations!

During the winter holiday, warm hearts and generosity can be felt near and far. We ask every patient and family to help us raise funds to continue our homocystinuria community outreach, education, and advocacy initiatives to educate providers and be there for families like Holly and Chris'. Please share our appeal letter with your colleagues, friends, and family.

Three Easy Ways to Donate

Text text the code

"HCU2024" to 44-321

By check
HCU Network America
15 S. Mallory Ave
Batavia, IL 60510

EMPLOYER MATCHING GIFT PROGRAM

Did you submit for your Employers Corporate Matching Gifts Program?

What are Matching Gifts?
Corporate matching gifts are a type of gift giving in which companies financially match donations that their employees make to nonprofit organizations. When an employee makes a donation, they need to request the matching gift from the employer, who then makes their own donation. Companies usually match at a 1:1 ratio, but some will match 1:2 or 2:1.

Why are Corporate Matching Gifts Valuable?
Corporate matching gifts are valuable because they are free money for your nonprofit of choice, HCU Network America! Your donation has double the power without you having to give double the amount.

Does HCU Network America Really Benefit?
If corporate matching gifts weren't valuable, we wouldn't be sending this letter. In 2020 we received approximately $45,000 in corporate matching gifts!

How Do I Find out if my Employer has a Corporate Matching Gifts Program?
Your company website or websites such as doublethedonation.com make it easy for you to search and see if your employer takes part in corporate matching gifts. If you can't find it online, please consult with your employee handbook, HR rep or manager to find out.

How Do I Request my Donation is Matched by my Employer?

1. The donor completes their donation
2. The donor submits matching gift request
3. Company reviews donation and nonprofit eligibility and reaches out to nonprofit
4. Nonprofit verifies the donation was made
5. If eligible, the nonprofit will receive the matching gifts request!

Top Matching Gift Companies

Company	Match Ratio
General Electric	1:1
Gap Corporation	1:1
ExxonMobil	3:1
Johnson & Johnson	2:1
Microsoft	1:1
Pfizer	1:1
Coca-Cola	2:1
And many more!	

Did you know some companies match retired employees donations?

What is Giving Tuesday?

It's a global generosity movement unleashing the power of radical generosity. GivingTuesday was created in 2012 as a simple idea: a day that encourages people to do good. Since then, it has grown into a year-round global movement that inspires hundreds of millions of people to give, collaborate, and celebrate generosity. *This year, Giving Tuesday will be* ***December 3.***

How do I get involved?

We're asking you to assist us in reaching our $10,000 GivingTuesday fundraising goal.

To get started, set up your own GivingTuesday fundraiser on GoFundMe Facebook, or Instagram! Setting up your fundraiser on these platforms is simple & HCU Network America receives 100% of the donations!

- Start to reach out to your friends and family in advance and get them to pledge a donation first thing on December 3rd!
- Let them know that by giving to HCU Network America, they are supporting programs and resources that directly benefit the patients and caregivers of our community.
- Remind them that *all donations* are tax-deductible AND will be matched by 3 anonymous donors (up to $25,000!)

How do I set up a GoFundMe fundraiser?

It's easy! Select 'charity' and type in HCU Network America. From there, just follow the prompts! GoFundMe is great because you can link in a video if you'd like. Consider a quick video telling how our programs have made a positive impact on you, along with your ask. Or, you can always post an image along with text.

GIVING TUESDAY

UPCOMING EVENTS

iGive.com

You Shop. Your Charity Gets Money. For Free.

Your holiday shopping makes a difference

Shop at any of the 1,700 stores that want to help, and **HCU Network America** will receive a portion of your purchase, ranging from .5% to over 20%! Stores pay for it all. Never pay more, and sometimes less with coupons and deals.

Want to make your purchases count?

The optional **iGive Button** is a simple web browser app, easy to install and uninstall. It automatically activates at participating stores.

Don't want the Button or an app? Just start your shopping trips by going to iGive.com.

Shop normally (no special codes, no special anything) at any of about 1,700 stores. The Button is working in the background to let them know you're helping when you shop.

Ready to shop?

Head to igive.com to sign up! Make your first purchase within 30 days and iGive will donate a bonus $5 to HCU Network America! Happy shopping!

Walmart · HSN · QVC · KOHL'S · L.L.Bean · macy's · 1-800-flowers.com · Booking.com · Clarks · GAP · Shipt · & more!

HC&U Podcast

Meet your hosts!

Welcome to the HC&U Podcast! We are Ben and Lindsey, your hosts. We are so excited to host this podcast as extra resource for the Homocystinuria community. We hope you like our content!

To Listen:

https://hcunetworkamerica.org/hcu-podcast/

or click below on your favorite option!

LISTEN ON Spotify

Listen on Apple Podcasts

LISTEN ON iHeartRADIO

Listen on amazon music

The latest episode

Ben chats with twin brothers Vicente & Victor, and Valerie, all adults living with Classical HCU. They'll talk growing up with HCU, Family planning, work, and advice to younger patients and families.

Bonus episode!

In this bonus episode, Ben chats with Kristen, a preclinical consultant focusing on rare diseases, about research and drug development, and potential new therapies for HCU.

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

POWERED BY RARE X

106 people have enrolled in the HCU Data Collection Program (DCP)

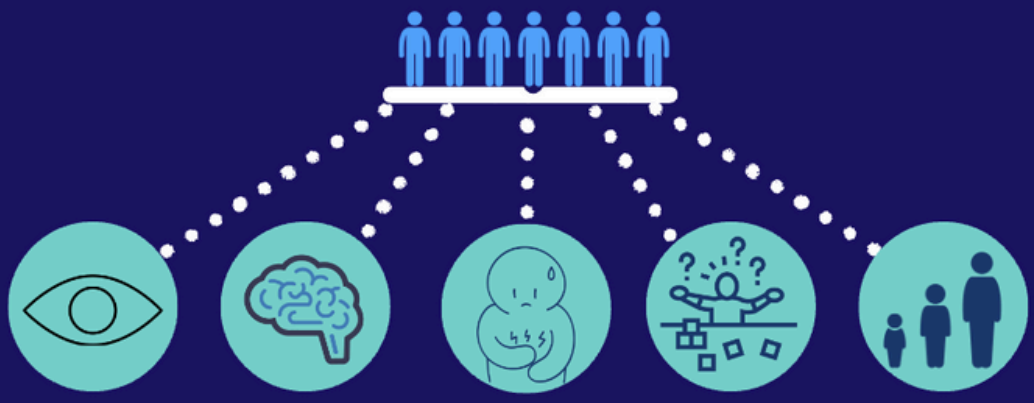
74 have Classical HCU

32 cobalamin disorder or MTHFR

Only 71 have completed the Head to Toe Survey

Without you completing this survey, your data is incomplete!

Of the 71 participants who took the Head-to-Toe Survey
here are the top 5 areas the data is pointing too



46 Vision issues

44 Central Nervous System issues

35 Digestive issues

32 Developmental Issues

32 Growth Issues

Complete the Head to Toe Survey at
https://rare-x.org/homocystinuria/

CANPCU+ CANPKU+ HCU

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HCU Network Australia

Now Enrolling

Acappella

Sponsor: Travere Therapeutics
Study type: Natural History (no investigational medicine given)
Study duration: About 6.5 years
Goal: To learn more about classical HCU & the course of the disease

TO QUALIFY*	AGE OF PARTICIPANTS	DETAILS
Diagnosis of Homocystinuria due to CBS Deficiency (Classical Homocystinuria)	Currently enrolling 1 to 4 years old	The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years.

**Your child will need to meet all other study criteria to take part in the ACAPPELLA Study.*

Study Locations
United States: Colorado, Washington DC, Georgia, Pennsylvania
Countries outside of the US: Ireland and Qatar

Approximately 150 people aged between 1 and 65 will participate at sites in the US, Europe and other countries around the world.

150

The ACAPPELLA Study has already enrolled 100 adults and children over 5 years old, and is now looking for children aged 1 to 4 years old to take part.

You may be able to receive payment for time and travel if your child participates in this study.

Talk to your doctor and family members about your child joining the ACAPPELLA study.

Sites are open and currently enrolling participants. For the most current information about the ACAPPELLA Study and to see additional eligibility criteria, please visit:

https://www.clinicaltrials.gov/study/NCT02998710

If you have any questions, please email:

medinfo@travere.com

For more information, please scan the QR code or visit:

www.hcuconnection.com.

MA-PE-24-0008. Version 2.0; en-US; March 2024

TRAVERE THERAPEUTICS

Sign our NBS Screening Petition!

In this petition, we call for state newborn screening labs to **revise screening protocols for Classical Homocystinuria** to *ensure fewer false negative screening results and delayed diagnoses.*

Click here to sign the petition!

Get your kit!

Our FREE Customizable Kits are here!

Request yours today!

At HCU Network America, we believe that one of the most important steps to empowering patients and caregivers is giving them the support and tools needed to succeed! We know that a new diagnosis can be overwhelming and riddled with concerns and questions. To us, one way to combat those feelings, and give you the confidence you need, is by providing you with one-on-one support, educational resources, and practical tools, such as scales, cooler bags, and more! Our request for a kit survey allows you the opportunity to request a one-on-one introductory call (with more opportunities to connect), and then a customized kit to the patient's needs. Don't want a call or a Zoom? That's fine too - we are happy to send you the customized kit.

Request your kit now - **https://www.surveymonkey.com/r/HCUKitSurvey**

**Kits can only be sent to patients in the continental US. However, we are happy to connect virtually and share the educational materials with you via weblinks*!

Stay connected: Join our Contact Register!

What is a contact register?

- a secured private survey that allows you to share information on you or your family member with HCU with us. (general contact info, diagnosis, etc)
- kept confidential and will not be shared unless you permit us

Why join?

- subscribe to our monthly newsletter and other communications
- identify other patients in your state and request their contact information
- access information posted over time that can only be shared with the patient community
- helps us plan events and inform the development of resources and educational tools
- supports our advocacy efforts and enables us to have informed conversations with doctors, pharmaceutical companies, state newborn screening labs, and lawmakers.

I want to participate! What's next?

You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to:
https://www.surveymonkey.com/r/HCUContact

HOPE CONNECTS US

FOLLOW US